

WHAT IS CLAIMED IS:

1. A method for screening for an increased risk of hypercalciuria comprising:

- (a) obtaining a sample nucleic acid from a subject; and
- (b) analyzing the sample nucleic acid to detect the presence or absence of a genetic mutation in genomic region associated with an increased risk of developing hypercalciuria.

2. The method of claim 1, wherein the hypercalciuria is further defined as absorptive hypercalciuria.

3. The method of claim 1, wherein the hypercalciuria is further defined as osteoporosis with hypercalciuria.

4. The method of claim 3, wherein the osteoporosis with hypercalciuria is further defined as ideopathic osteoporosis with hypercalciuria.

5. The method of claim 3, wherein the osteoporosis with hypercalciuria is further defined as postmenopausal osteoporosis with hypercalciuria.

6. The method of claim 1, wherein the nucleic acid is DNA.

7. The method of claim 1, wherein the subject is a human.

8. The method of claim 7, wherein genomic region is comprised in chromosome 1.

9. The method of claim 8, wherein the genomic region is comprised in 1q23 and 1q24.

10. The method of claim 9, wherein the genomic region is located between markers D1S2681 and D1S2815.

11. The method of claim 9, wherein the genomic region has a sequence contained in SEQ ID NO:1.

12. The method of claim 1, wherein the genomic region has a sequence contained in at least one genetic sequence selected from the group consisting of the the genetic sequences set forth in GenBank Accession # Z97876, GenBank Accession # Z99943, and GenBank Accession # AL031733.

13. The method of claim 1, wherein the genomic region has a lod score of greater than 3.0 but less than 30.0.

14. The method of claim 1, wherein analyzing the sample nucleic acid is done with a PCR procedure, diagnostic RFLP analysis, RNase protection assay, or RNase mismatch cleavage assay.

15. The method of claim 14, wherein analyzing the sample nucleic acid is done with a PCR procedure.

16. A method of treating hypercalciuria comprising
(a) screening for an increased risk of hypercalciuria using a sample nucleic acid from a patient;
(b) detecting said increased risk of hypercalciuria; and
(c) treating the patient with an increased risk of hypercalciuria.

17. The method of claim 16, wherein the screening for an increased risk of hypercalciuria comprises:

(a) obtaining a sample nucleic acid from a subject; and
(b) analyzing the sample nucleic acid to detect the presence or absence of a genetic mutation in genomic region associated with an increased risk of developing hypercalciuria.

18. The method of claim 16, wherein the hypercalciuria is further defined as absorptive hypercalciuria.

19. The method of claim 16, wherein the hypercalciuria is further defined as osteoporosis with hypercalciuria.

20. The method of claim 19, wherein the osteoporosis with hypercalciuria is further defined as ideopathic osteoporosis with hypercalciuria.

21. The method of claim 19, wherein the osteoporosis with hypercalciuria is further defined as postmenopausal osteoporosis with hypercalciuria

22. The method of claim 16, wherein treating the patient comprises the use of therapeutic, dietary or fluid regimens.

23. The method of claim 22, wherein the treating the patient further comprises increased fluid intake, restricted sodium intake or increased animal protein intake.

24. The method of claim 22, wherein the treating the patient further comprises the use of thiazides, sodium cellulose phosphate therapy, or slow-release, neutral potassium phosphate salt.

25. The method of claim 22, wherein the treating the patient further comprises the treatment to prevent stone formation.

26. A method for familial screening for an increased risk of absorptive hypercalciuria or osteoporosis with hypercalciuria comprising:

(a) obtaining a sample nucleic acid from a patient;

(b) analyzing the sample nucleic acid to detect the presence of a marker known to be linked genetically to region of human chromosome 1, wherein the region is associated with an increased risk of developing absorptive hypercalciuria.

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